AGXT gene

alanine--glyoxylate aminotransferase

Normal Function

The *AGXT* gene provides instructions for making an enzyme called alanine-glyoxylate aminotransferase. This enzyme is found in liver cells, specifically within cell structures called peroxisomes. These structures are important for several cellular activities, such as ridding the cell of toxic substances and helping to break down certain fats. In the peroxisome, alanine-glyoxylate aminotransferase converts a compound called glyoxylate to the protein building block (amino acid) glycine.

Health Conditions Related to Genetic Changes

Primary hyperoxaluria

More than 175 mutations in the *AGXT* gene have been found to cause primary hyperoxaluria type 1. This condition is caused by the overproduction of a substance called oxalate. Excess amounts of this substance lead to kidney and bladder stones, which can begin anytime from childhood to early adulthood with kidney disease developing at any age. Deposition of oxalate in multiple other tissues throughout the body (systemic oxalosis) can cause additional health problems.

Most of the *AGXT* gene mutations decrease or eliminate alanine-glyoxylate aminotransferase activity, which impairs the conversion of glyoxylate to glycine. Other mutations cause the enzyme to be misplaced in cells, transporting it to structures called mitochondria instead of to peroxisomes. While the enzyme in the mitochondria retains activity, it cannot access glyoxylate, which is in peroxisomes. All *AGXT* gene mutations result in the accumulation of glyoxylate, which is converted to oxalate instead of glycine. The oxalate is filtered through the kidneys and is either excreted in urine as a waste product or combines with calcium to form calcium oxalate, a hard compound that is the main component of kidney and bladder stones. Increased oxalate levels in the blood can lead to systemic oxalosis, particularly affecting bones and the walls of blood vessels in people with primary hyperoxaluria type 1.

Other Names for This Gene

- AGT
- AGT1

- AGXT1
- alanine glyoxylate aminotransferase
- alanine-glyoxylate aminotransferase
- alanine-glyoxylate aminotransferase (oxalosis I; hyperoxaluria I; glycolicaciduria; serine-pyruvate aminotransferase)
- alanine-glyoxylate transaminase
- L-alanine: glyoxylate aminotransferase 1
- pyruvate (glyoxylate) aminotransferase
- serine-pyruvate aminotransferase
- serine:pyruvate aminotransferase
- SPAT
- SPT

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of AGXT (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=189[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28AGXT%5BTIAB%5D%29+OR+%28alanine-glyoxylate+aminotransferase%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- ALANINE-GLYOXYLATE AMINOTRANSFERASE; AGXT (https://omim.org/entry/604285)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/189)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=AGXT[gene])

References

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Genomic Location

The *AGXT* gene is found on chromosome 2 (https://medlineplus.gov/genetics/chromosome/2/).

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